O10-1

Enlarging the genetic spectrum of cardiomyopathies. Preliminary results of the MyEstela Pediatric Cohort Study.

Medrano-López C.(1), Vázquez-Álvarez M.C.(1), Camino-López M. (1), Blanco-Soto P. (1), Centeno-Jiménez M. (1), Monserrat-Iglesias L. (2), Orera-Clemente M. (1), Maroto-Álvaro E. (1) The Myestela Study Group
Hospital General Universitario Gregorio Marañón, Madrid, Spain (1); Complexo Hospitalario Universitario de A Coruña, A Coruña, Spain (2)

Introduction: The knowledge of the genetic origin of pediatric cardiomyopathies has increased over the last years. Dilated cardiomyopathy (DCM) is the most common form, Hypertrophic cardiomyopathy (HCM) and Left ventricular noncompaction (LVNC) are commonly inherited as an autosomal dominant trait. The objective of this study is to describe the genetic origin of a cohort of pediatric patients with cardiomyopathy.

Methods: Prospective observational cohort study from June to December 2010 of children followed up in a familial cardiopathies outpatients clinic diagnosed with idiopathic cardiomyopathy. Demographics, family tree and genetic test for protein-coding sequences of sarcomere protein genes (MYH7, MYBPC3, TNNI3, TNNT2, TPM1, MYL2, MYL3, ACTC, and TNNC1) were performed.

Results: 36 patients (44,4% male) were included in the cohort. 26 genetic results have been received and 10 (38.5%) were positive. 20% (1/5) were positive in the group under 1 year of age, 28,6% (4/14) in the group between 1 and 10 years and 71,4% (5/7) in the group over 10 years. 40% of the positive results were male.

	Num patients	Gender	Age (years) Mean(range)	Positive results	Gene type/ num positives
DCM	15	7M/8F	3,6(0-14,4)	2/13(15,4%)	2/2
HCM	9	6M/3F	9,3(0,2-16,5	5/6(83,3%)	5/5
LVNC	12	3M/9F	8(1,6-14,3)	3/7(42,9%)	1/3
Total	36	16M/20F	6,5 (0-16,5)	10/26(38,5%)	5/10

The mutations were found in 5 of the 9 genes studied: 5 MYH7 (1 HCM, 1 DCM, 3 LVNC), 2 TNNT (1 HCM, 1 DCM), 1 ACTC (HCM), 1MyBPC3 (HCM), 1 MyL3 (HCM). 7 of them were novel mutations, not previously described in literature (3 HCM, 2 DCM, 2LVNC). At present 3 families have been studied, 1 being a de novo mutation and 2 an inherited mutation. During the study period two patients with HCM were implanted with a Cardioverter Defibrillator , two DCM died, one infant of sudden death and a new born with a prenatal hydrops diagnosis.

Conclusions: Mutations in cardiac sarcomere proteins are a common cause of cardiomyopathy in our initial cohort of children, more frequently in the oldest group and in those with HCM and LVNC. The description of high rate of novel mutations will improve the knowledge on pediatric cardiomyopathies.